



TBP gene

TATA-box binding protein

Normal Function

The *TBP* gene provides instructions for making a protein called the TATA box binding protein. This protein is active in cells and tissues throughout the body, where it plays an essential role in regulating the activity of most genes.

The TATA box binding protein attaches (binds) to a particular sequence of DNA known as the TATA box. This sequence occurs in a regulatory region of DNA near the beginning of many genes. Once the protein is attached to the TATA box near a gene, it acts as a landmark to indicate where other enzymes should start reading the gene. The process of reading a gene's DNA and transferring the information to a similar molecule called mRNA is known as transcription.

One region of the *TBP* gene contains a particular DNA segment known as a CAG/CAA trinucleotide repeat. This segment is made up of a series of three DNA building blocks (nucleotides) that appear multiple times in a row. Normally, the CAG/CAA segment is repeated 25 to 42 times within the gene.

Health Conditions Related to Genetic Changes

Huntington disease-like syndrome

A particular type of mutation in the *TBP* gene has been found to cause a progressive brain disorder known as Huntington disease-like 4 (HDL4) or spinocerebellar ataxia type 17 (SCA17). The features of this disorder vary widely among affected individuals. The condition was first described as HDL4 in people whose signs and symptoms closely resembled those of Huntington disease, including uncontrolled movements, emotional problems, and loss of thinking ability. The disorder is now more commonly known as SCA17 because difficulty coordinating movements (ataxia) and other movement problems are the most frequent signs and symptoms. It is unknown why some people with *TBP* mutations have a disorder resembling Huntington disease, while others have more prominent ataxia.

The mutation associated with HDL4/SCA17 increases the size of the CAG/CAA trinucleotide repeat in the *TBP* gene. People with this condition have 43 to 66 CAG/CAA repeats. People with 43 to 48 CAG/CAA repeats may or may not have signs and symptoms, while people with 49 or more repeats almost always develop the disorder.

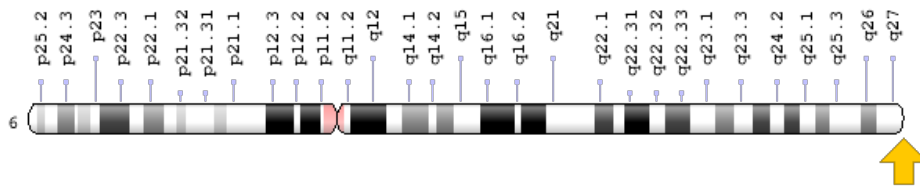
An increased number of CAG/CAA repeats in the *TBP* gene leads to the production of an abnormally long version of the TATA box binding protein. The abnormal protein

builds up in nerve cells (neurons) in the brain and disrupts the normal functions of these cells. The dysfunction and eventual death of neurons in certain areas of the brain underlie the signs and symptoms of HDL4/SCA17. Because the *TBP* gene is active throughout the body, it is unclear why the effects of a mutation in this gene are limited to the brain.

Chromosomal Location

Cytogenetic Location: 6q27, which is the long (q) arm of chromosome 6 at position 27

Molecular Location: base pairs 170,554,333 to 170,572,870 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CCG1 Protein
- CCGS
- Cell Cycle Gene 1 Protein
- DYT3 protein, human
- GTF2D
- GTF2D1
- RNA Polymerase II TATA-Binding Protein
- RNA Polymerase IIA 250kD
- SCA17
- TAF(II)250
- TAF1 RNA Polymerase II TATA Box Binding Protein
- TAF2A
- TAFII250
- TATA-Binding Protein

- TATA box binding protein
- TATA-Box Factor
- TATA Sequence-Binding Protein
- TBP_HUMAN
- TF2D
- TFIID
- Transcription Factor IID
- Transcription Factor TBP
- Transcription Initiation Factor TFIID 250 kDa Subunit

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The TATA-Box-Binding Protein Initiates the Assembly of the Active Transcription Complex
<https://www.ncbi.nlm.nih.gov/books/NBK22433/#A3988>
- Protein Data Bank Molecule of the Month: TATA-Binding Protein
<http://pdb101.rcsb.org/motm/67>

GeneReviews

- Spinocerebellar Ataxia Type 17
<https://www.ncbi.nlm.nih.gov/books/NBK1438>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TATA+box+binding+protein%5BMAJR%5D%29+OR+%28SCA17%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

OMIM

- TATA BOX-BINDING PROTEIN
<http://omim.org/entry/600075>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TBP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TBP%5Bgene%5D>
- HGNC Gene Family: General transcription factors
<http://www.genenames.org/cgi-bin/genefamilies/set/565>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11588
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6908>
- UniProt
<http://www.uniprot.org/uniprot/P20226>

Sources for This Summary

- Bauer P, Laccone F, Rolfs A, Wüllner U, Bösch S, Peters H, Liebscher S, Scheible M, Epplen JT, Weber BH, Holinski-Feder E, Weirich-Schwaiger H, Morris-Rosendahl DJ, Andrich J, Riess O. Trinucleotide repeat expansion in SCA17/TBP in white patients with Huntington's disease-like phenotype. *J Med Genet.* 2004 Mar;41(3):230-2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/14985389>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735701/>
- Gao R, Matsuura T, Coolbaugh M, Zühlke C, Nakamura K, Rasmussen A, Siciliano MJ, Ashizawa T, Lin X. Instability of expanded CAG/CAA repeats in spinocerebellar ataxia type 17. *Eur J Hum Genet.* 2008 Feb;16(2):215-22. Epub 2007 Nov 28.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18043721>
- Rasmussen A, De Biase I, Fragoso-Benítez M, Macías-Flores MA, Yescas P, Ochoa A, Ashizawa T, Alonso ME, Bidichandani SI. Anticipation and intergenerational repeat instability in spinocerebellar ataxia type 17. *Ann Neurol.* 2007 Jun;61(6):607-10.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17474109>
- Rolfs A, Koeppen AH, Bauer I, Bauer P, Buhlmann S, Topka H, Schöls L, Riess O. Clinical features and neuropathology of autosomal dominant spinocerebellar ataxia (SCA17). *Ann Neurol.* 2003 Sep; 54(3):367-75.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12953269>
- Schneider SA, van de Warrenburg BP, Hughes TD, Davis M, Sweeney M, Wood N, Quinn NP, Bhatia KP. Phenotypic homogeneity of the Huntington disease-like presentation in a SCA17 family. *Neurology.* 2006 Nov 14;67(9):1701-3.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17101913>

- Zühlke C, Hellenbroich Y, Dalski A, Kononowa N, Hagenah J, Vieregge P, Riess O, Klein C, Schwinger E. Different types of repeat expansion in the TATA-binding protein gene are associated with a new form of inherited ataxia. *Eur J Hum Genet.* 2001 Mar;9(3):160-4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11313753>
 - van Roon-Mom WM, Reid SJ, Faull RL, Snell RG. TATA-binding protein in neurodegenerative disease. *Neuroscience.* 2005;133(4):863-72. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15916858>
-

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/TBP>

Reviewed: August 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services